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CASE REPORT

Scleral Cyst Associated with Anomalous Tilted Configuration of the Optic Nerve Head: A Case Report

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ABSTRACT

There is limited published data currently available on scleral cysts in the posterior pole. Here, we detail the case of a patient who was suspected to have a peripapillary scleral cyst imprinting the optic nerve head (ONH) profile. The 52-year-old asymptomatic and otherwise healthy woman presented with unilateral ONH blurred margins of the left eye for 1 year. Her best-corrected visual acuity in the right and left eye was 20/20 and 20/25, respectively. Fundus observation of the right eye revealed no significant abnormalities; on the left eye a tilted disc with blurred margins in the superior quadrants and gliosis-associated changes in the lower quadrants were identified. Structural optical coherence tomography (OCT) showed a posterior hyporeflective cystic space at the level of the ONH, and OCT-angiography revealed flow void. The differential diagnosis of ONH edema was considered and the case discussed with the neuro-ophthalmology unit. Given the clinical history, the absence of symptoms and the multimodal imaging findings, a peripapillary scleral cyst was considered to be the most likely explanation for the edematous appearance and the anomalous tilted configuration of the ONH. This case suggests that although rare, even more so in the absence of an ONH coloboma, a postequatorial scleral cyst should be considered in the differential diagnosis of ONH lesions.

Keywords: Ocular coherence tomography; Ocular coherence tomography angiography; Optic nerve; Scleral cyst

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INTRODUCTION

Epithelial cysts of the sclera are seldom reported and when reported they are most often encountered in the anterior sclera, with or without cornea involvement [1–4]. A few studies have reported the increased incidence of retrobulbar cysts in patients with cavitary anomalies of the optic nerve head (ONH), such as a coloboma and morning glory syndrome [5]. Recently, a case of postequatorial scleral cyst

mimicking a choroid melanoma was published in which the cyst was located in the superotemporal quadrant of the retina [6]. Here, we present and discuss a case of a woman affected by a scleral cyst located beneath the superior part of the ONH.

METHODS

The EQUATOR network CARE guidelines [7] for case reports were followed. The study protocol complied with the tenets of the Declaration of Helsinki, and informed consent was obtained from the patient for her case to be presented and discussed.

The clinical records of the patient were reviewed, including demographic data, best-corrected visual acuity (BCVA) and results from the spectral-domain optical coherence tomography (OCT) (Spectralis & HRA; Heidelberg Engineering, Heidelberg, Germany) and OCT-Angiography (PLEX® Elite 9000; Carl Zeiss Meditec Inc., Dublin, CA, USA) examinations.

CASE REPORT

A 52-year-old woman was referred to the Medical Retina & Imaging Unit of our department after a screening ophthalmological examination 1 year earlier had identified blurred margins of the ONH in the left eye. With the exception of a family history of age-related macular degeneration, she had no other relevant past conditions and was otherwise healthy. She denied any subjective complaints regarding changes in recent visual acuity or other associated symptoms.

Her BCVA was 20/20 (refraction − 0.75 diopters [D]) in the right eye and 20/25 (refraction − 1.50 D) in the left eye. Intraocular pressure was 16 mmHg in both eyes, and the results of the anterior segment and vitreous examination were unremarkable. Axial length was 26.0 and 26.5 mm in the right and left eye, respectively. Fundus observation of the right eye revealed no significant abnormalities, but in the left eye, a tessellated and irregular fundus was observed, along with a tilted disc with blurred margins in the superior quadrants and gliosis-associated changes in the lower quadrants (Fig. 1a). No other overlying changes of abnormal aspects were observed in the remaining part of the retina.

As depicted in Fig. 1, infrared reflectance imaging examination of the ONH and of the posterior pole was normal. The cross-sectional analysis using structural spectral-domain OCT revealed an elevation of the superior quadrant of the ONH with a posterior hyporeflective round space (Fig. 1a, b). Swept-Source OCT-angiography of the same region was performed,

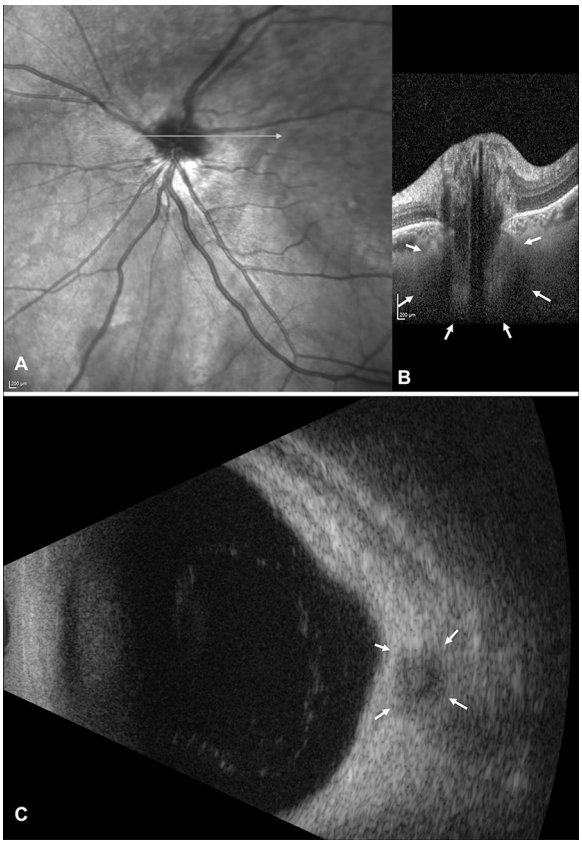

Fig. 1 **a**, **b** Infrared reflectance image of the optic nerve head (ONH) and of the peripapillary retina (**a**) and corresponding structural spectral-domain ocular coherence tomography (OCT) B-scan crossing the ONH of the left eye (**b**). A tilted disk appearance is denoted, and a well-delimited, approximately round structure beneath the ONH is observable (white arrows). **c** Ultrasonography showed a slight elevation of the ONH, along with an hypoechogenic round structure below the optic disk (white arrows)

and custom segmentation of the area of interest revealed a void of flow and tissue that was compatible with a cyst (Fig. 2). The ultrasonography revealed a slight elevation of the ONH, along with a hypoechogenic round structure below the optic disk (Fig. 1c). Visual field examination showed slight enlargement of the blind spot in the left eye, overlapping with the findings of the examination performed 1 year earlier. Orbital and brain magnetic resonance imaging were performed and the results were unremarkable.

DISCUSSION

Several cases of retrobulbar cysts have been reported in the literature, but these were associated with trauma or ocular surgery [5]. In our case, there was no history of trauma or ocular surgery, and the retrobulbar cyst was detected in absence of ONH coloboma but in presence of a tilted disc appearance. Tilted disc syndrome is a congenital anomaly of the eye that could range from a minimal situs inversus of the retinal vessels to a full coloboma of the inferior fundus [8]. Tilted disc syndrome could manifest as different visual field defects, of which the most common is an upper temporal field defect directed towards the blind spot [9]. The diagnosis of this syndrome is very important in clinical practice because the visual field defects associated with tilted disc syndrome can be confused with chiasmal lesions or glaucoma defects [8]. The clinical and imaging features of our patients (i.e. a tilted disc appearance at fundus examination and ultrasonography; a slight enlargement of the blind spot on the visual field examination; and the mismatch between the manifest refraction and the axial length) confirmed the diagnosis of tilted disc syndrome. Furthermore, multimodal imaging approach allowed us to identify a cystic space located beneath the ONH that caused blurring of its superior quadrants' contour, which motivated the referral to our center. The differential diagnosis of ONH edema was considered, and the case was discussed with the neuro-ophthalmology unit of our department. Given the clinical history, the absence of symptoms, the unremarkable results of the orbital and brain magnetic resonance imaging and the multimodal imaging findings, an optic neuritis or other cause of ONH edema was excluded. A peripapillary scleral cyst beneath the ONH was considered to be the most likely explanation for the edematous appearance and the anomalous tilted configuration of the ONH.

As the current hypotheses for the appearance of scleral cysts in the anterior segment [1, 3] and in the context of an ONH coloboma [5] are not applicable to this case, further research may

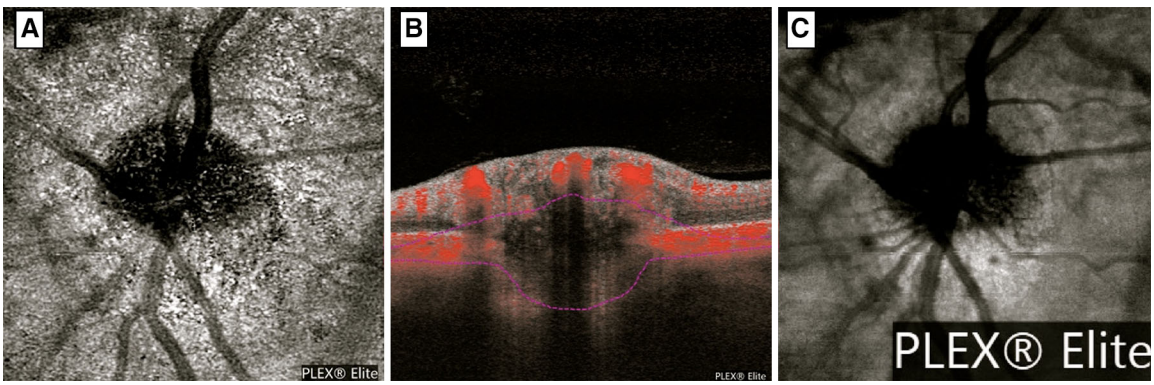

Fig. 2 Optical coherence tomography angiography (OCT-A) of the left optic nerve head (**a** 3 × 3-mm en-face OCT-A image with custom segmentation, **b** structural cross-sectional B-scan with flow showing the custom segmentation boundaries, **c** 3 × 3-mm en-face structural OCT with the same segmentation as illustrated in **a**. The ONH vessels are pushed forward, noted as areas with blood flow on the B-scan. A flow and tissue void space is noted beneath the ONH

help enlighten the origin of posterior pole scleral cysts. Also, serial follow-up of this case would allow us to understand if these cysts are likely to remain stable or to change its dimensions over time. In this context, the peripapillary gliosis of the inferior quadrants may suggest that the cyst could have been larger, causing further changes in the ONH and a consequent remodeling of the surrounding tissues. To the contrary, the absence of vision complaints or other associated symptoms and the asymptomatic diagnosis may suggest that the changes to this cyst have been occurring very slowly.

Although this is the first reported case of a peripapillary scleral cyst, it should be noted that current advances in imaging would certainly improve our diagnostic ability to detect similar cases in the future.

CONCLUSIONS

In conclusion, in this case report we discuss a rare differential diagnosis when blurred margins of the ONH are encountered on fundus examination of an asymptomatic patient. This work raises awareness of the importance of multimodal imaging and a correct diagnosis before potentially harmful treatments are considered. Although rare, even more so in the absence of an ONH coloboma, a postequatorial scleral cyst should be considered in the differential diagnosis of ONH lesions.

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Compliance with Ethics Guidelines. Informed consent was obtained from the individual participant for being included in the study.

Data Availability. Data sharing is not applicable to this article as no datasets were generated or analyzed during the current study.

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